

CLAIMS

What is claimed is:

- 5 1. An isolated and purified nucleic acid comprising a sequence encoding a protein selected from the group consisting of SEQ ID NOs: 2, 6, 8, 10, 12, 14, 16, and 18.
2. The nucleic acid sequence of Claim 1, wherein said sequence is operably
10 linked to a heterologous promoter.
3. The nucleic acid sequence of Claim 1, wherein said sequence is contained
 within a vector.
4. The nucleic acid sequence of Claim 3, wherein said vector is within a host
15 cell.
5. A kit comprising a reagent for detecting the presence or absence of a
 variant MCFD2 polypeptide in a biological sample.
- 20 6. The kit of claim 5, further comprising instruction for using said kit for said
 detecting the presence or absence of a variant MCFD2 polypeptide in a biological
 sample.
- 25 7. The kit of claim 5, further comprising instructions for diagnosing
 combined deficiency of factor V and factor VIII in said subject based on the presence or
 absence of said variant MCFD2 polypeptide.
8. The kit of claim 5, wherein said reagent is one or more antibodies.
- 30 9. The kit of claim 8, wherein said antibodies comprise a first antibody that
 specifically binds to the C-terminus of said MCFD2 polypeptide and a second antibody
 that specifically binds to the N-terminus of said MCFD2 polypeptide.
- 35 10. The kit of claim 9, wherein said variant MCFD2 polypeptide is a C-
 terminal truncation of SEQ ID NO:2.

11. The kit of claim 5, wherein said reagents are configured to detect a MCFD2 nucleic acid sequence.

5 12. A method for detection of a variant MCFD2 polypeptide in a subject, comprising:

a) providing a biological sample from a subject, wherein said biological sample comprises a MCFD2 polypeptide; and

10 b) detecting the presence or absence of a variant MCFD2 polypeptide in said biological sample.

13. The method of claim 12, wherein said variant MCFD2 polypeptide is a C-terminal truncation of SEQ ID NO:2.

15 14. The method of claim 12, wherein the presence of said variant MCFD2 polypeptide is indicative of combined deficiency of factor V and factor VIII in said subject.

20 15. The method of claim 12, wherein said biological sample is selected from the group consisting of a blood sample, a tissue sample, a urine sample, and an amniotic fluid sample.

25 16. The method of claim 12, wherein said subject is selected from the group consisting of an embryo, a fetus, a newborn animal, and a young animal.

17. The method of claim 12, wherein said detecting comprises differential antibody binding.

30 18. The method of claim 12, wherein said detecting comprises a gel-free truncation test.

19. The method of claim 12, wherein said detection comprises a Western blot.

35 20. The method of claim 12, wherein said detecting comprises detecting a MCFD2 nucleic acid sequence.